ABSTRACT

Lafora's disease in humans is characterized by the mutation or deletion of an *EPM2A* gene, which encodes a protein, Laforin, having a tyrosine phosphatase domain. Many different sequence mutations, including microdeletions, in *EPM2A* co-segregate with Lafora's disease. Accordingly, detection of mutations in *EPM2A* are useful in diagnosing Lafora's disease. – –